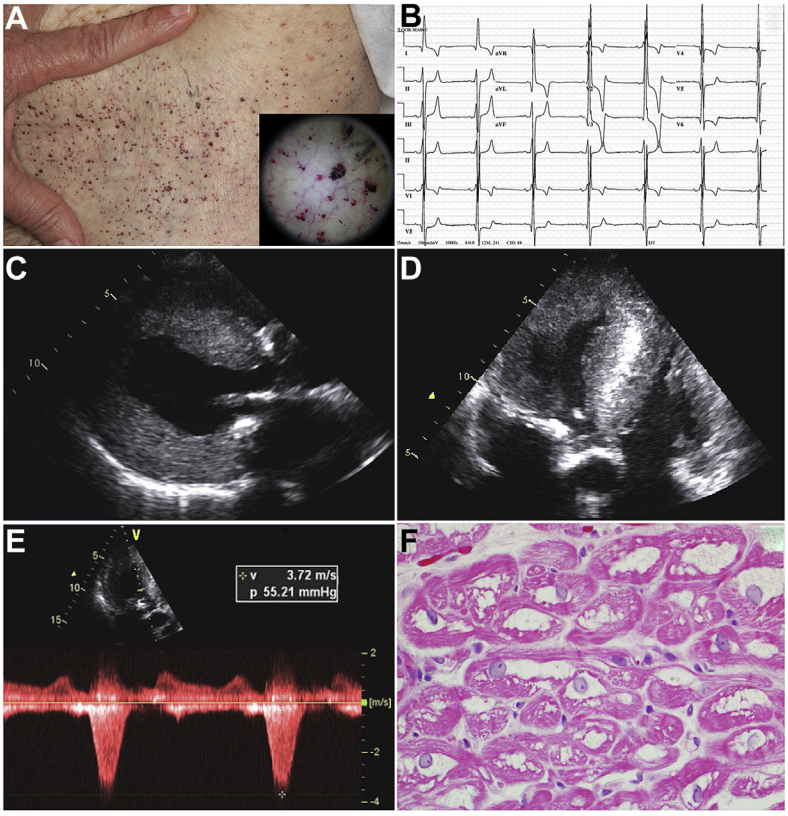
Journal of the American College of Cardiology
© 2014 by the American College of Cardiology Foundation
Published by Elsevier Inc.
Vol. 63, No. 17, 2014
ISSN 0735-1097/$36.00
http://dx.doi.org/10.1016/j.jacc.2013.07.121

IMAGES IN CARDIOLOGY

Fabry Disease With Resting Outflow Obstruction Masquerading as Hypertrophic Cardiomyopathy

Jeffrey B. Geske, MD,* Hayan Jouni, MD,* Marie C. Aubry, MD,† Bernard J. Gersh, MB, ChB, DPhil*

Rochester, Minnesota



From the *Division of Cardiovascular Diseases, Department of Internal Medicine, Mayo Clinic, Rochester, Minnesota; and the †Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, Minnesota. Dr. Gersh served as a consultant for PPD Development, Cardiovascular Research Foundation, Inspire-MD, Boston Scientific, Baxter Healthcare Corp., Medtronic Inc., and St. Jude Medical. All other authors have reported that they have no relationships relevant to this paper to disclose.

Manuscript received July 11, 2013; accepted July 30, 2013.

A 67-year-old man with prior coronary artery bypass grafting, rheumatic fever with mitral valve repair, and unexplained stage IV chronic kidney disease was referred for evaluation of obstructive hypertrophic cardiomyopathy because of progressive exertional dyspnea. Physical examination showed angiokeratomas **(A)**. Electrocardiography showed sinus bradycardia, right bundle branch block, left anterior fascicular block, and high voltages with anterolateral T-wave changes **(B)**. Transthoracic echocardiography revealed an ejection fraction of 59%, right and concentric left ventricular hypertrophy, and a resting left ventricular outflow gradient of 55 mm Hg (**C to E**, Online Videos 1 and 2). The patient's rash, striking electrocardiogram, right ventricle involvement, and renal failure raised suspicion of an alternate diagnosis despite resting obstruction. The patient's serum alpha-galactosidase level was 0.0 nmol/h/mg (normal >23.1 nmol/h/mg), and examination of a right ventricular biopsy specimen revealed myocyte hypertrophy with cytoplasmic vacuolization **(F)**, confirming Fabry disease. Differentiating Fabry disease from hypertrophic cardiomyopathy is crucial given the therapeutic and prognostic differences. The patient is currently undergoing evaluation for enzyme replacement therapy.